



Rhode Island Commission on Women

Position Paper on Genetics¹

The Rhode Island Commission on Women (RICW) supports universal, affordable, accessible, high quality, confidential health care for all women, as stated in its general position paper on women's health.² This care includes services related to the practice of medical genetics, which is the 24th medical specialty under the American Board of Medical Specialties.³ In addition, safeguards must be in place to protect patient confidentiality and the privacy of genetic information.

As stated in previous position papers, the RICW supports a woman's right to participate actively in decisions affecting her health and the health of her children. Meaningful participation requires an environment that supports autonomy, confidentiality, access to accurate, scientifically up-to-date and culturally appropriate information, and the opportunity to choose a comprehensive course of treatment and a qualified provider as measured by objective criteria and information provided by government public health sources. The RICW's position paper on the quality of care⁴ defines high quality health care as care that is accessible, effective, safe, accountable, culturally competent and fair. The paper further defines and expands the scope of this care. Medical genetics is a critical area of health care based on which life-altering decisions may be made. It is the RICW's position that access to comprehensive, confidential, culturally appropriate quality genetic services should be available to all Rhode Island women and their families. It is also the position of the RICW that access to genetic counseling is part of the access to the spectrum of genetic services, administered before testing to assist women in deciding if testing is right for them, and after testing to aid women in interpreting test results.⁵ As more and more genes are being discovered through the efforts of researchers participating in the Human Genome Project, many ethical, legal and social implications (ELSI) issues are being, and will be, raised in connection with genetic screening for the presence of these genes in certain individuals, diagnostic genetic testing, and therapeutic procedures. The testing of women for the presence of two breast cancer-related genes (i.e., BRCA1 and BRCA2) is an example. Unfortunately, many women currently lack adequate counseling prior to and after receiving genetic services.⁶ The Rhode Island Commission on Women believes that women should have better access to accurate and unbiased information about various genetic tests, including possible social, psychological, medical and economic impacts the results may have on themselves and their families.

In connection with genetic testing, the RICW believes that all genetic tests, including "home brews", must meet federal minimum standards before being approved and used outside of a research setting. These minimum standards must include the analysis of data regarding analytic and clinical validity and clinical utility to determine if a test is ready for clinical application. Each intended use of a test must be validated. A particularly stringent level of scrutiny for genetic tests that predict life threatening, chronic or disabling diseases must be applied. Genetic tests that fall under this category must be available only if and when the validity of the test is fully established. Furthermore, the Food and Drug Administration (FDA) must use its current regulatory authority to increase oversight of all genetic tests. Inconsistent, unreliable pre-market review of genetic tests creates the potential for great harm in a clinical setting. Without FDA review, tests are released prematurely, before information about their predictive value is available. To make informed health decisions, individuals must be assured that the tests provide reliable, accurate information through FDA oversight for the protection of human subjects participating in all genetic test research including "home-brew" genetic tests.

The RICW recognizes the right of consumers to access high quality genetic services through providers with the proper experience and credentials (e.g., licensure and board certification in the appropriate specialty of genetics or medicine) and by facilities with the proper accreditation.^{7, 8, 9} The RICW supports policies and legislation that ensure the delivery of high quality genetic services to all stakeholders. Furthermore, all stakeholders should be granted ready access to information on genetic testing facilities and personnel.

Ethical, legal, and social implication (ELSI) issues inherent to genetic testing must be fully addressed to prevent abuse. For example, there is great pressure to utilize new genetic technologies developed in the laboratory (see, for example, Mark, Miller and Jenkins, 1994)¹⁰ despite limited knowledge of potential risks and benefits, or for conditions for which there is no known effective treatment. Thus, various interrelated issues, such as autonomy, informed consent, veracity, utility, non-maleficence, personal dignity, beneficence, the right to access the latest genetic technology, patient confidentiality and justice, costs and benefits, have been raised in connection with genetic testing.¹¹ Additional analyses of opportunities and threats posed by genetic knowledge and technology will need to be performed by society as a whole. There are also risks of employment and insurance discrimination.^{12, 13, 14} The American College of Medical Genetics (ACMG), in a recent position paper¹⁵, stated that fear of genetic discrimination in employment and health insurance may have a negative impact on consumers' willingness to seek genetic services and to participate in genetic research. This behavior will impede access to health care for women and their families. Therefore, the RICW supports the enforcement of legislation for preventing genetic discrimination, such as a recently enacted bill that would make genetic discrimination by insurance companies illegal in Rhode Island.¹⁶ Safeguards must be in place to protect patient confidentiality and the privacy of genetic information. Towards this end, the Health Insurance Portability and Accountability Act of 1996 (the HIPAA privacy rules) which created new requirements for health care providers to protect the privacy and security of health information, became effective April 14, 2001 and mandated that all health care providers must be in compliance with the Rules by April 14, 2003.¹⁷ Questions such as who should be tested and/or be covered for testing, and issues such as the commercialization and marketing of genetic services, will continue to be raised in the ongoing national genetic debates and will continue to be explored.

The RICW recognizes the urgent need for more genetics education for many audiences, brought on by the advent of the Human Genome Project and the sequencing of the human genome^{18, 19} and for funding to determine the exact content, scope, scale and methods of delivery/ implementation in Rhode Island. As President Bush noted²⁰, we are living in a time of accelerating medical progress. With this information comes enormous possibilities for doing good. Through a better understanding of medical genetics, including its limitations, scientists might one day be able to cure and prevent genetic diseases.

We must insure that the public is protected and that all genetic tests meet rigorous scientific standards, regulated and enforced by the federal government. In summary, the RICW supports appropriate representation of women of diverse cultural, racial, ethnic, and social backgrounds in our national research agenda especially when it pertains to issues related to the human genome, prevention, and treatment of genetic diseases, so as to fulfill America's passion for scientific excellence in genetic knowledge and technology, and its vision for a just society with equality and fairness for all people with diverse phenotypes and genotypes.

GLOSSARY OF TERMS

Analytic validity: assuring that the lab performance of a genetic test is accurate and precise.

Autonomy: self-rule or personal self-governance: personal rule of the self by adequate understanding while remaining free from controlling interferences by others and from personal limitations that prevent choice. Autonomy is the freedom from external constraint and the presence of critical mental capacities such as understanding, planning, and deciding.²¹

Beneficence: (doing of good and the active promotion of good, kindness, and charity). Beneficence requires us to abstain from injuring others and to help others further their important and legitimate interests, largely by preventing or removing possible harms.²²

Clinical validity: refers to several measures of clinical performance including clinical sensitivity and clinical specificity, and also positive predictive values. It refers to genotype-phenotype correlations.

Clinical utility: refers to how information from the test can be used in a clinical context to improve people's health, whether the benefits outweigh the risks.

Home brew: refers to genetic tests developed by the laboratory itself.

Informed Consent: a physician's obligation to disclose information to the quality of a patient's or subject's understanding of information and the right of a patient to authorize or refuse a biomedical intervention. Informed consent has the following elements: disclosure, comprehension, voluntariness, competence, and consent.²³

Non-maleficence: not to inflict evil or harm.

Veracity: to tell the truth

Utility: the greatest good or happiness for the greatest number.

¹ Approved by the RICW Health Committee on January 28, 2002, approved as amended by the Executive Committee on March 25, 2002, and approved with amendments by the Full Commission on April 1, 2002.

² Rhode Island Commission on Women, 2000. *Position Paper on Women's Health*. Adopted by the Rhode Island Commission on Women on January 11, 2000.

³ Mark, H.F.L. and R.E. Pyeritz, 2000. *Cytogenetics as a Specialty of Medical Genetics*. In Mark, H.F.L. (editor). Medical Cytogenetics, New York: Marcel Dekker, Inc.

⁴ Rhode Island Commission on Women, 2001. *Position Paper on Quality of Health Care*. Adopted by the Rhode Island Commission on Women on March 12, 2001.

⁵ Koenig, B.A., Greely, H.T., McConnell, L.M., Silverberg, H.L., Raffin, T.A., and the members of the Breast Cancer Working Group of the Stanford Program in Genomics, Ethics and Society, 1998. *Genetic Testing for BRCA1 and BRCA2: recommendations of the Stanford Program in Genomics, Ethics and Society*. Journal of Women's Health 7 (5): 531-545.

⁶ McCance, Kathryn L. and Lynn B. Jorde, 1998. *Evaluating the Genetic Risk of Breast Cancer*. The Nurse Practitioner.

⁷ Mark, H.F.L., et al., 1995. *Current Issues of Personnel and Laboratory Practices in Genetic Testing*. J. Medical Genetics 32: 780-786.

⁸ Mark, H.F.L. and M. Watson, 1994. *Evolving Standards of Practice for Clinical Cytogenetics*. Rhode Island Medicine 77: 373-374.

⁹ Williams, M.S., 2001. *Genetics and Managed Care: Policy Statement of the American College of Medical Genetics*. Genetics in Medicine 3(6): 430-435.

¹⁰ Mark, H.F.L., W. Miller and R. Jenkins, 1996. *Current Applications of Molecular Cytogenetics Technologies*. Annals Clin. Lab. Science 27:47-56.

¹¹ Alexakos, Fran, 2001. *Rhode Island Primary Care Physicians Attitudes to Ordering Genetic Testing for Breast Cancer*. Salve Regina University, Newport, RI.

¹² Council for Responsible Genetics, 2001. *Genetic Discrimination Position Paper*. www.genewatch.org/progrmas/GD_PP.

¹³ Matloff, E.T., H. Shappell, K. Brierley, B.A. Bernhardt, W. McKinnon, and B.N. Peshkin, 2000. *What would you do? Specialists' perspective on cancer genetic testing*. J Clin Oncol 18 (12) 2484-2492.

¹⁴ Watson, M.S. and C.L. Greene, 2001. *Points to Consider in Preventing Unfair Discrimination Based on Genetic Disease Risk: A Position Statement of the American College of Medical Genetics*. Genetics in Medicine 3(6): 430-435.

¹⁵ American College of Medical Genetics, 2001. *Points to Consider in Preventing Unfair Discrimination Based on Genetic Disease Risk*. American College of Medical Genetics, Bethesda, MD.

¹⁶ S 0803 SUBSTITUTE A, 2001. *An Act Relating to Insurance – Genetic Testing*. Rhode Island General Assembly.

¹⁷ Fleisher, L.D., and L.J. Cole, 2001. *Health Insurance Portability and Accountability is Here: What Price Privacy?* Genetics in Medicine 3, No. 4: 286-289.

¹⁸ International Human Genome Sequencing Consortium, 2001. *Initial Sequencing and Analysis of the Human Genome*. Nature 409: 860-921.

¹⁹ Venter CJ, et al., 2001. *The Human Genome*. Science 291: 1304-1351.

²⁰ Bush, G.W., 2001. *President George W. Bush's Radio Address to the Nation*. Saturday, June 23, 2001.

²¹ Beauchamp, Tom L., LeRoy Walters. *Contemporary Issues in Bioethics*. 4th ed. Belmont: Wardsworth Press, 1994.

²² Ibid.

²³ Ibid.